Hirotomo Saitsu

List of Publications by Citations

199 papers	4,632 citations	36 h-index	62 g-index
234 ext. papers	5,818 ext. citations	4.9 avg, IF	4.96 L-index

#	Paper	IF	Citations
199	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. *Nature Genetics*, **2008**, 40, 782-8	36.3	416
198	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. *Nature Genetics*, **2012**, 44, 376-8	36.3	350
197	De novo mutations in the autophagy gene WDR45 cause static encephalopathy of childhood with neurodegeneration in adulthood. *Nature Genetics*, **2013**, 45, 445-9, 449e1	36.3	330
196	Human genetic variation database, a reference database of genetic variations in the Japanese population. *Journal of Human Genetics*, **2016**, 61, 547-53	4.3	212
195	De Novo mutations in GNAO1, encoding a Gβ subunit of heterotrimeric G proteins, cause epileptic encephalopathy. *American Journal of Human Genetics*, **2013**, 93, 496-505	11	137
194	Mutations in POLR3A and POLR3B encoding RNA Polymerase III subunits cause an autosomal-recessive hypomyelinating leukoencephalopathy. *American Journal of Human Genetics*, **2011**, 89, 644-51	11	112
193	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. *Neurogenetics*, **2013**, 14, 225-32	3	91
192	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. *Cell Reports*, **2018**, 22, 734-747	10.6	86
191	DNA methylation and gene expression dynamics during spermatogonial stem cell differentiation in the early postnatal mouse testis. *BMC Genomics*, **2015**, 16, 624	4.5	84
190	De novo SOX11 mutations cause Coffin-Siris syndrome. *Nature Communications*, **2014**, 5, 4011	17.4	84
189	Whole exome sequencing identifies KCNQ2 mutations in Ohtahara syndrome. *Annals of Neurology*, **2012**, 72, 298-300	9.4	74
188	Phenotypic spectrum of GNAO1 variants: epileptic encephalopathy to involuntary movements with severe developmental delay. *European Journal of Human Genetics*, **2016**, 24, 129-34	5.3	72
187	De novo mutations in SLC35A2 encoding a UDP-galactose transporter cause early-onset epileptic encephalopathy. *Human Mutation*, **2013**, 34, 1708-14	4.7	72
186	Spatial and temporal expression of folate-binding protein 1 (Fbp1) is closely associated with anterior neural tube closure in mice. *Developmental Dynamics*, **2003**, 226, 112-7	2.9	72
185	The somatic GNAQ mutation c.548G>A (p.R183Q) is consistently found in Sturge-Weber syndrome. *Journal of Human Genetics*, **2014**, 59, 691-3	4.3	70
184	De novo KCNT1 mutations in early-onset epileptic encephalopathy. *Epilepsia*, **2015**, 56, e121-8	6.4	66
183	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. *Epilepsia*, **2013**, 54, 1262-9	6.4	64

182	Biallelic Mutations in MYPN, Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. *American Journal of Human Genetics*, **2017**, 100, 169-178	11	57
181	GRIN1 mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. *Epilepsia*, **2015**, 56, 841-8	6.4	56
180	Ineffective quinidine therapy in early onset epileptic encephalopathy with KCNT1 mutation. *Annals of Neurology*, **2016**, 79, 502-3	9.4	52
179	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. *Scientific Reports*, **2016**, 6, 30072	4.9	52
178	Mutations in genes encoding polycomb repressive complex 2 subunits cause Weaver syndrome. *Human Mutation*, **2017**, 38, 637-648	4.7	50
177	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. *Brain*, **2018**, 141, 1703-1718	11.2	44
176	De novo WDR45 mutation in a patient showing clinically Rett syndrome with childhood iron deposition in brain. *Journal of Human Genetics*, **2014**, 59, 292-5	4.3	44
175	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. *Brain*, **2017**, 140, 2322-2336	11.2	44
174	Biallelic mutations in the 3Pexonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. *Nature Genetics*, **2017**, 49, 457-464	36.3	43
173	High prevalence of genetic alterations in early-onset epileptic encephalopathies associated with infantile movement disorders. *Brain and Development*, **2016**, 38, 285-92	2.2	42
172	PARS2 and NARS2 mutations in infantile-onset neurodegenerative disorder. *Journal of Human Genetics*, **2017**, 62, 525-529	4.3	41
171	Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an Pexome-firstPapproach. *Journal of Human Genetics*, **2015**, 60, 175-82	4.3	41
170	Precise detection of chromosomal translocation or inversion breakpoints by whole-genome sequencing. *Journal of Human Genetics*, **2014**, 59, 649-54	4.3	40
169	Expanding the phenotypic spectrum of TUBB4A-associated hypomyelinating leukoencephalopathies. *Neurology*, **2014**, 82, 2230-7	6.5	38
168	De novo KIF1A mutations cause intellectual deficit, cerebellar atrophy, lower limb spasticity and visual disturbance. *Journal of Human Genetics*, **2015**, 60, 739-42	4.3	37
167	De novo hotspot variants in CYFIP2 cause early-onset epileptic encephalopathy. *Annals of Neurology*, **2018**, 83, 794-806	9.4	37
166	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism. *Human Genetics*, **2018**, 137, 95-104	6.3	36
165	Quinidine therapy for West syndrome with KCNTI mutation: A case report. *Brain and Development*, **2017**, 39, 80-83	2.2	36

164	WDR45 mutations in three male patients with West syndrome. *Journal of Human Genetics*, **2016**, 61, 653-61	4.3	36
163	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. *Scientific Reports*, **2017**, 7, 3552	4.9	35
162	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 950-961	11	34
161	A case of autism spectrum disorder arising from a de novo missense mutation in POGZ. *Journal of Human Genetics*, **2015**, 60, 277-9	4.3	33
160	ANKRD11 variants cause variable clinical features associated with KBG syndrome and Coffin-Siris-like syndrome. *Journal of Human Genetics*, **2017**, 62, 741-746	4.3	31
159	De novo KCNH1 mutations in four patients with syndromic developmental delay, hypotonia and seizures. *Journal of Human Genetics*, **2016**, 61, 381-7	4.3	30
158	Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures. *Journal of Human Genetics*, **2019**, 64, 313-322	4.3	29
157	variants in and cause neurodevelopmental disorders. *Annals of Clinical and Translational Neurology*, **2018**, 5, 280-296	5.3	27
156	Two cases of early-onset myoclonic seizures with continuous parietal delta activity caused by EEF1A2 mutations. *Brain and Development*, **2016**, 38, 520-4	2.2	27
155	Early infantile epileptic encephalopathy associated with the disrupted gene encoding Slit-Robo Rho GTPase activating protein 2 (SRGAP2). *American Journal of Medical Genetics, Part A*, **2012**, 158A, 199-205	2.5	25
154	Bortezomib-resistance is associated with increased levels of proteasome subunits and apoptosis-avoidance. *Oncotarget*, **2016**, 7, 77622-77634	3.3	25
153	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. *Journal of Human Genetics*, **2016**, 61, 527-31	4.3	24
152	Characteristic MRI findings in beta-propeller protein-associated neurodegeneration (BPAN). *Neurology: Clinical Practice*, **2014**, 4, 175-177	1.7	24
151	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. *Journal of Human Genetics*, **2016**, 61, 835-8	4.3	24
150	De novo IGF2 mutation on the paternal allele in a patient with Silver-Russell syndrome and ectrodactyly. *Human Mutation*, **2017**, 38, 953-958	4.7	23
149	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. *Human Molecular Genetics*, **2018**, 27, 1421-1433	5.6	23
148	Paternal germline mosaicism of a SCN2A mutation results in Ohtahara syndrome in half siblings. *European Journal of Paediatric Neurology*, **2014**, 18, 567-71	3.8	23
147	A case of early onset epileptic encephalopathy with de novo mutation in SLC35A2: Clinical features and treatment for epilepsy. *Brain and Development*, **2017**, 39, 256-260	2.2	23

146	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. *Nature Communications*, **2019**, 10, 2506	17.4	22
145	De novo missense mutations in NALCN cause developmental and intellectual impairment with hypotonia. *Journal of Human Genetics*, **2016**, 61, 451-5	4.3	22
144	Molecular Diagnosis of 34 Japanese Families with Leber Congenital Amaurosis Using Targeted Next Generation Sequencing. *Scientific Reports*, **2018**, 8, 8279	4.9	21
143	De novo SHANK3 mutation causes Rett syndrome-like phenotype in a female patient. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1593-6	2.5	21
142	mutations cause variable phenotypes of developmental and epileptic encephalopathy. *Epilepsia Open*, **2018**, 3, 495-502	4	21
141	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. *Journal of Medical Genetics*, **2019**, 56, 396-407	5.8	20
140	Biallelic COLGALT1 variants are associated with cerebral small vessel disease. *Annals of Neurology*, **2018**, 84, 843-853	9.4	20
139	Identification of novel SNORD118 mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. *Clinical Genetics*, **2017**, 92, 180-187	4	19
138	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. *Human Mutation*, **2017**, 38, 1542-1554	4.7	19
137	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. *European Journal of Paediatric Neurology*, **2016**, 20, 412-7	3.8	19
136	Three Cases of KCNT1 Mutations: Malignant Migrating Partial Seizures in Infancy with Massive Systemic to Pulmonary Collateral Arteries. *Journal of Pediatrics*, **2017**, 191, 270-274	3.6	17
135	Epileptic apnea in a patient with inherited glycosylphosphatidylinositol anchor deficiency and PIGT mutations. *Brain and Development*, **2018**, 40, 53-57	2.2	17
134	Different patterns of cerebellar abnormality and hypomyelination between POLR3A and POLR3B mutations. *Brain and Development*, **2014**, 36, 259-63	2.2	17
133	The first report of Japanese patients with asparagine synthetase deficiency. *Brain and Development* , **2017**, 39, 236-242	2.2	17
132	De novo DNM1 mutations in two cases of epileptic encephalopathy. *Epilepsia*, **2016**, 57, e18-23	6.4	17
131	RNA sequencing solved the most common but unrecognized NEB pathogenic variant in Japanese nemaline myopathy. *Genetics in Medicine*, **2019**, 21, 1629-1638	8.1	17
130	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. *Scientific Reports*, **2014**, 4, 7132	4.9	16
129	Ataxic phenotype with altered Ca3.1 channel property in a mouse model for spinocerebellar ataxia 42. *Neurobiology of Disease*, **2019**, 130, 104516	7.5	15

128	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. *Journal of Human Genetics*, **2018**, 63, 263-270	4.3	15
127	Severe leukoencephalopathy with cortical involvement and peripheral neuropathy due to FOLR1 deficiency. *Brain and Development*, **2017**, 39, 266-270	2.2	15
126	A novel WDR45 mutation in a patient with static encephalopathy of childhood with neurodegeneration in adulthood (SENDA). *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2388-90	2.5	15
125	A hemizygous GYG2 mutation and Leigh syndrome: a possible link?. *Human Genetics*, **2014**, 133, 225-34	6.3	15
124	Novel compound heterozygous LIAS mutations cause glycine encephalopathy. *Journal of Human Genetics*, **2015**, 60, 631-5	4.3	14
123	De novo variants in RHOBTB2, an atypical Rho GTPase gene, cause epileptic encephalopathy. *Human Mutation*, **2018**, 39, 1070-1075	4.7	14
122	A novel mutation in the proteolytic domain of LONP1 causes atypical CODAS syndrome. *Journal of Human Genetics*, **2017**, 62, 653-655	4.3	13
121	De novo variants in cause intellectual disability, autism spectrum disorder, and epilepsy with myoclonic absences. *Epilepsia Open*, **2019**, 4, 476-481	4	13
120	Germline-Derived Gain-of-Function Variants of Gs-Coding Gene Identified in Nephrogenic Syndrome of Inappropriate Antidiuresis. *Journal of the American Society of Nephrology: JASN*, **2019**, 30, 877-889	12.7	13
119	IGF2 Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	13
118	MYRF haploinsufficiency causes 46,XY and 46,XX disorders of sex development: bioinformatics consideration. *Human Molecular Genetics*, **2019**, 28, 2319-2329	5.6	12
117	Electroclinical features of epileptic encephalopathy caused by SCN8A mutation. *Pediatrics International*, **2015**, 57, 758-62	1.2	12
116	De novo PHACTR1 mutations in West syndrome and their pathophysiological effects. *Brain*, **2018**, 141, 3098-3114	11.2	12
115	Neuroimaging findings in Joubert syndrome with C5orf42 gene mutations: A milder form of molar tooth sign and vermian hypoplasia. *Journal of the Neurological Sciences*, **2017**, 376, 7-12	3.2	11
114	Exome reports A de novo GNB2 variant associated with global developmental delay, intellectual disability, and dysmorphic features. *European Journal of Medical Genetics*, **2020**, 63, 103804	2.6	11
113	Prenatal clinical manifestations in individuals with variants. *Journal of Medical Genetics*, **2021**, 58, 505-513	3.8	11
112	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. *Journal of Human Genetics*, **2018**, 63, 1223-1229	4.3	11
111	A severe pulmonary complication in a patient with COL4A1-related disorder: A case report. *European Journal of Medical Genetics*, **2017**, 60, 169-171	2.6	10

110	De Novo Truncating Variants in the Last Exon of SEMA6B Cause Progressive Myoclonic Epilepsy. *American Journal of Human Genetics*, **2020**, 106, 549-558	11	10
109	Biallelic Variants in CNPY3, Encoding an Endoplasmic Reticulum Chaperone, Cause Early-Onset Epileptic Encephalopathy. *American Journal of Human Genetics*, **2018**, 102, 321-329	11	10
108	Cerebellar ataxia-dominant phenotype in patients with ERCC4 mutations. *Journal of Human Genetics*, **2018**, 63, 417-423	4.3	10
107	De novo HDAC8 mutation causes Rett-related disorder with distinctive facial features and multiple congenital anomalies. *Brain and Development*, **2018**, 40, 406-409	2.2	10
106	Compound heterozygous GFM2 mutations with Leigh syndrome complicated by arthrogryposis multiplex congenita. *Journal of Human Genetics*, **2015**, 60, 509-13	4.3	10
105	A Japanese case of cerebellar ataxia, spastic paraparesis and deep sensory impairment associated with a novel homozygous TTC19 mutation. *Journal of Human Genetics*, **2015**, 60, 187-91	4.3	10
104	Involvement of the axially condensed tail bud mesenchyme in normal and abnormal human posterior neural tube development. *Congenital Anomalies (discontinued)*, **2008**, 48, 1-6	1.1	10
103	A female case of aromatic l-amino acid decarboxylase deficiency responsive to MAO-B inhibition. *Brain and Development*, **2016**, 38, 959-963	2.2	10
102	GRIN2D variants in three cases of developmental and epileptic encephalopathy. *Clinical Genetics*, **2018**, 94, 538-547	4	10
101	POLR3A variants in striatal involvement without diffuse hypomyelination. *Brain and Development*, **2020**, 42, 363-368	2.2	9
100	Partial androgen insensitivity syndrome caused by a deep intronic mutation creating an alternative splice acceptor site of the AR gene. *Scientific Reports*, **2018**, 8, 2287	4.9	9
99	Two novel homozygous RAB3GAP1 mutations cause Warburg micro syndrome. *Human Genome Variation*, **2015**, 2, 15034	1.8	9
98	Identification of a novel homozygous SPG7 mutation in a Japanese patient with spastic ataxia: making an efficient diagnosis using exome sequencing for autosomal recessive cerebellar ataxia and spastic paraplegia. *Internal Medicine*, **2013**, 52, 1629-33	1.1	9
97	A de novo variant in RAC3 causes severe global developmental delay and a middle interhemispheric variant of holoprosencephaly. *Journal of Human Genetics*, **2019**, 64, 1127-1132	4.3	8
96	Identification of a deep intronic POLR3A variant causing inclusion of a pseudoexon derived from an Alu element in Pol III-related leukodystrophy. *Journal of Human Genetics*, **2020**, 65, 921-925	4.3	8
95	Clinical characteristics of a Japanese patient with Bardet-Biedl syndrome caused by BBS10 mutations. *Japanese Journal of Ophthalmology*, **2018**, 62, 458-466	2.6	8
94	Two Japanese cases of epileptic encephalopathy associated with an FGF12 mutation. *Brain and Development*, **2018**, 40, 728-732	2.2	8
93	Recurrent SCN3A p.Ile875Thr variant in patients with polymicrogyria. *Annals of Neurology*, **2018**, 84, 159-161	9.4	8

92	ATP6V0A1 encoding the a1-subunit of the V0 domain of vacuolar H-ATPases is essential for brain development in humans and mice. *Nature Communications*, **2021**, 12, 2107	17.4	8
91	A case of de novo splice site variant in SLC35A2 showing developmental delays, spastic paraplegia, and delayed myelination. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e814	2.3	7
90	A male case with CDKL5-associated encephalopathy manifesting transient methylmalonic acidemia. *European Journal of Medical Genetics*, **2018**, 61, 451-454	2.6	7
89	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. *Journal of Human Genetics*, **2017**, 62, 997-1000	4.3	7
88	PCortical cerebellar atrophyPdwindles away in the era of next-generation sequencing. *Journal of Human Genetics*, **2014**, 59, 589-90	4.3	7
87	Quinidine therapy and therapeutic drug monitoring in four patients with KCNT1 mutations **2019**, 21, 48-54		7
86	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. *Journal of Human Genetics*, **2021**, 66, 401-407	4.3	7
85	A de novo TOP2B variant associated with global developmental delay and autism spectrum disorder. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1145	2.3	6
84	Long-term observation of a Japanese mucolipidosis IV patient with a novel homozygous p.F313del variant of MCOLN1. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1500-1505	2.5	6
83	A case of new PCDH12 gene variants presented as dyskinetic cerebral palsy with epilepsy. *Journal of Human Genetics*, **2018**, 63, 749-753	4.3	6
82	Deletions of SCN2A and SCN3A genes in a patient with West syndrome and autistic spectrum disorder. *Seizure: the Journal of the British Epilepsy Association*, **2018**, 60, 91-93	3.2	6
81	A recurrent homozygous variant in siblings with Lafora disease. *Human Genome Variation*, **2018**, 5, 16	1.8	6
80	A case of tubulinopathy presenting with porencephaly caused by a novel missense mutation in the TUBA1A gene. *Brain and Development*, **2018**, 40, 819-823	2.2	6
79	Comprehensive clinical and molecular studies in split-hand/foot malformation: identification of two plausible candidate genes (LRP6 and UBA2). *European Journal of Human Genetics*, **2019**, 27, 1845-1857	5.3	6
78	De novo truncating variants in PHF21A cause intellectual disability and craniofacial anomalies. *European Journal of Human Genetics*, **2019**, 27, 378-383	5.3	6
77	Identification of novel compound heterozygous mutations in ACO2 in a patient with progressive cerebral and cerebellar atrophy. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00698	2.3	5
76	De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms. *Journal of Human Genetics*, **2020**, 65, 727-734	4.3	5
75	Folate receptors and neural tube closure. *Congenital Anomalies (discontinued)*, **2017**, 57, 130-133	1.1	4

74	CCNB2 and AURKA overexpression may cause atypical mitosis in Japanese cortisol-producing adrenocortical carcinoma with TP53 somatic variant. *PLoS ONE*, **2020**, 15, e0231665	3.7	4
73	Nonsense-associated altered splicing of MAP3K1 in two siblings with 46,XY disorders of sex development. *Scientific Reports*, **2020**, 10, 17375	4.9	4
72	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. *Human Genetics*, **2021**, 140, 1109-1120	6.3	4
71	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. *Human Mutation*, **2021**, 42, 66-76	4.7	4
70	Insulin resistant diabetes mellitus in SHORT syndrome: case report and literature review. *Endocrine Journal*, **2021**, 68, 111-117	2.9	4
69	Foxc2 knock-in mice mark stage-specific Foxc2-expressing cells during mouse organogenesis. *Congenital Anomalies (discontinued)*, **2017**, 57, 24-31	1.1	3
68	A novel homozygous truncating variant of NECAP1 in early infantile epileptic encephalopathy: the second case report of EIEE21. *Journal of Human Genetics*, **2019**, 64, 347-350	4.3	3
67	Different types of suppression-burst patterns in patients with epilepsy of infancy with migrating focal seizures (EIMFS). *Seizure: the Journal of the British Epilepsy Association*, **2019**, 65, 118-123	3.2	3
66	A case of early-onset epileptic encephalopathy with a homozygous TBC1D24 variant caused by uniparental isodisomy. *American Journal of Medical Genetics, Part A*, **2019**, 179, 645-649	2.5	3
65	Coexistence of a CAV3 mutation and a DMD deletion in a family with complex muscular diseases. *Brain and Development*, **2019**, 41, 474-479	2.2	3
64	Early-onset epileptic encephalopathy and severe developmental delay in an association with de novo double mutations in and. *Epilepsia Open*, **2018**, 3, 81-85	4	3
63	A de novo GABRB2 variant associated with myoclonic status epilepticus and rhythmic high-amplitude delta with superimposed (poly) spikes (RHADS). *Epileptic Disorders*, **2020**, 22, 476-481	1.9	3
62	Nanopore sequencing reveals a structural alteration of mirror-image duplicated genes in a genome-editing mouse line. *Congenital Anomalies (discontinued)*, **2020**, 60, 120-125	1.1	3
61	Genetic and phenotypic analysis of 101 patients with developmental delay or intellectual disability using whole-exome sequencing. *Clinical Genetics*, **2021**, 100, 40-50	4	3
60	De novo ATP1A3 variants cause polymicrogyria. *Science Advances*, **2021**, 7,	14.3	3
59	Clinical manifestations and epilepsy treatment in Japanese patients with pathogenic CDKL5 variants. *Brain and Development*, **2021**, 43, 505-514	2.2	3
58	ZNF445: a homozygous truncating variant in a patient with Temple syndrome and multilocus imprinting disturbance. *Clinical Epigenetics*, **2021**, 13, 119	7.7	3
57	Expanding the concept of peroxisomal diseases and efficient diagnostic system in Japan. *Journal of Human Genetics*, **2019**, 64, 145-152	4.3	3

56	Dystonia due to bilateral caudate hemorrhage associated with a COL4A1 mutation. *Parkinsonism and Related Disorders*, **2017**, 40, 80-82	3.6	2
55	A patient with Muenke syndrome manifesting migrating neonatal seizures. *Brain and Development*, **2017**, 39, 873-876	2.2	2
54	A Male Case with Propeller Protein-Associated Neurodegeneration (BPAN) with Somatic Mosaic Mutation in WDR45 **2016**, 6,		2
53	A recurrent TMEM106B mutation in hypomyelinating leukodystrophy: A rapid diagnostic assay. *Brain and Development*, **2020**, 42, 603-606	2.2	2
52	De novo AFF3 variant in a patient with mesomelic dysplasia with foot malformation. *Journal of Human Genetics*, **2019**, 64, 1041-1044	4.3	2
51	De novo mutations in epilepsy. *Developmental Medicine and Child Neurology*, **2011**, 53, 806-807	3.3	2
50	De novo ZBTB7A variant in a patient with macrocephaly, intellectual disability, and sleep apnea: implications for the phenotypic development in 19p13.3 microdeletions. *Journal of Human Genetics*, **2020**, 65, 181-186	4.3	2
49	Myoclonic tremor status as a presenting symptom of adenylosuccinate lyase deficiency. *European Journal of Medical Genetics*, **2020**, 63, 104061	2.6	2
48	Life-threatening muscle complications of COL4A1-related disorder. *Brain and Development*, **2020**, 42, 93-97	2.2	2
47	Low-prevalence mosaicism of chromosome 18q distal deletion identified by exome-based copy number profiling in a child with cerebral hypomyelination. *Congenital Anomalies (discontinued)*, **2020**, 60, 94-96	1.1	2
46	Familial periodic paralysis associated with a rare KCNJ5 variant that supposed to have incomplete penetrance. *Brain and Development*, **2021**, 43, 470-474	2.2	2
45	Primary ovarian insufficiency in a female with phosphomannomutase-2 gene (PMM2) mutations for congenital disorder of glycosylation. *Endocrine Journal*, **2021**, 68, 605-611	2.9	2
44	Leigh syndrome-like MRI changes in a patient with biallelic variants treated with ketogenic diet. *Molecular Genetics and Metabolism Reports*, **2021**, 29, 100800	1.8	2
43	TSC1 intragenic deletion transmitted from a mosaic father to two siblings with cardiac rhabdomyomas: Identification of two aberrant transcripts. *European Journal of Medical Genetics*, **2020**, 63, 104060	2.6	1
42	A case of childhood glaucoma with a combined partial monosomy 6p25 and partial trisomy 18p11 due to an unbalanced translocation. *Ophthalmic Genetics*, **2020**, 41, 175-182	1.2	1
41	Single-fiber electromyography-based diagnosis of CACNA1A mutation in children: A potential role of the electrodiagnosis in the era of whole exome sequencing. *Brain and Development*, **2019**, 41, 905-909	2.2	1
40	Retinitis pigmentosa with optic neuropathy and mutations: A case report.. *American Journal of Ophthalmology Case Reports*, **2022**, 25, 101298	1.3	1
39	An intronic GNAO1 variant leading to in-frame insertion cause movement disorder controlled by deep brain stimulation.. *Neurogenetics*, **2022**, 23, 129	3	1

38	Compound heterozygous ADAMTS9 variants in Joubert syndrome-related disorders without renal manifestation. *Brain and Development*, **2021**,	2.2	1
37	Retrotransposition disrupting EBP in a girl and her mother with X-linked dominant chondrodysplasia punctata.. *Journal of Human Genetics*, **2022**,	4.3	1
36	Novel Variants Cause Progressive Leukodystrophy in Childhood: Case Report and Literature Review. *Child Neurology Open*, **2021**, 8, 2329048X211048613	1.3	1
35	De novo CACNA1G variants in developmental delay and early-onset epileptic encephalopathies. *Journal of the Neurological Sciences*, **2020**, 416, 117047	3.2	1
34	GNAO1 organizes the cytoskeletal remodeling and firing of developing neurons. *FASEB Journal*, **2020**, 34, 16601-16621	0.9	1
33	Parthenogenetic mosaicism: generation via second polar body retention and unmasking of a likely causative PER2 variant for hypersomnia. *Clinical Epigenetics*, **2021**, 13, 73	7.7	1
32	Comprehensive genetic analysis confers high diagnostic yield in 16 Japanese patients with corpus callosum anomalies. *Journal of Human Genetics*, **2021**, 66, 1061-1068	4.3	1
31	Cerebrovascular diseases in two patients with entire NSD1 deletion. *Human Genome Variation*, **2021**, 8, 20	1.8	1
30	Identification of two novel de novo TUBB variants in cases with brain malformations: case reports and literature review. *Journal of Human Genetics*, **2021**, 66, 1193-1197	4.3	1
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